Research Paper Medical Sciences

A Case of Skeletal Cystic Angiomatosis, A-V Malformations, and Arnold- Chiari Malformation Type I – A Rare Multicentric Combination Triad of Congenital Malformations.

Dr. A. Ramya	Intern (House-surgeon), Government Siddhartha Medical College, Vijayawada-520008, Andhra Pradesh.
Dr. K. V. Seshaiah, M.D.,	Assistant Professor of Medicine, Government Siddhartha Medical College, Vijayawada-520008, Andhra Pradesh.
Dr. K. Ramalakshmi, M.D.,	Professor & HOD of Medicine, ACSR Government Medical College, Nellore-524002, Andhra Pradesh

ABSTRACT

A 17-year-old male patient was admitted into our medical ward with complaints of progressive overgrowth of jawbone and occiput since 10 years; protrusion of eyeballs since 3 years; recent onset of weakness in all 4 limbs, paraesthesias, headache, neckpain, and slurred speech. Clinical evaluation revealed small pulsatile swelling over tongue and kyphoscoliosis. Neuro-imaging revealed cystic lesions in facial bones, calvarium and multiple vertebrae; arterio-venous malformations of brain; enlarged intra-orbital veins; and chiari malformation type-I. Biopsy from mandible confirmed the diagnosis of skeletal cystic angiomatosis. Finally an unusual combination triad of skeletal cystic angiomatosis, arterio-venous malformations, and chiari malformation type-I was documented in our patient. This rare, multi-centric combination triad of congenital malformations could not be found in literature.

KEYWORDS Arnold chiari malformation, Arterio-venous malformation, Cerebral haemorrhage, Cystic angiomatosis, Magnetic-resonance imaging.

Introduction

The term cystic angiomatosis (C A) was introduced in1953 by Jacobs and Kimmelstiel. The term 'C A' is preferred by most, to designate a pathologic entity that involves both the vascular and lymphatic systems, producing diffuse cystic lesions in both the axial and appendicular skeleton, with or without visceral involvement. Literature revealed reporting of only one case with combination of Arnold - chiari malformations and CA[1]; and only two cases with combination of CA and Rendu-osler-weber syndrome[2]. But inspite of vigorous search of literature, we could not find the unusual combination triad of CA, A-V malformations, and chiari malformation,as seen in our patient.

Case Report

A 17-year-old male, born of consanguinous marriage, was admitted into our medical ward with the complaints of progressive overgrowth of the jawbone and occiput since 10 years; protrusion of both eyeballs since 3 years; and progressive weakness of all 4 limbs since 1 month (Fig.1). Bony swellings were painless and he was afebrile. He also complained of neck pain; diffuse headache; paraesthesias; and slurred speech. Bowels and micturition were normal. His birth was normal and had normal milestones of development. He had normal intelligence and completed his secondary school education. Parents denied any history of trauma, seizures or childhood infections. He was non-hypertensive and non-diabetic.

Fig1: Photograph of the Patient

On examination, abnormal findings include prominence of jaw and occiput; bilateral exophthalmos; kyphoscoliosis; greying of hair; and pulsatile swelling over right side of tongue. Neurological evaluation revealed bilateral wasting of thenar and hypothenar muscles; bilateral wrist drop; increased tone in left upper and lower limbs; decreased muscle power of grade 3-4 in all four limbs; and dysarthria. Patient was conscious and coherent with normal higher intellectual functions. Clinical evaluation of Cardiovascular, Respiratory and Abdomen were unremarkable.

Hematology, biochemistry including serum calcium,and thyroid profile were unremarkable. Chest x-ray, ECG, echocardiogram and ultrasonography of abdomen were normal.

Plain computerised tomography (CT) scan of brain showed prominent ventricles with asymmetrical dilatation of left lateral ventricle; thickening of calvarium and facial bones with few lytic lesions; and destruction of left zygomatic bone and left parietal bone. Repeated CT scan of brain after 2 days revealed

haemorrhage into brain parenchyma of left temporo-parietal region, when patient developed sudden right-sided hemiplegia during his hospital stay. Magnetic resonance imaging (MRI) of brain showed parenchymal bleed in left temporal lobe; expanded skull bones with multiple cystic areas suggestive of cystic angiomatosis; and multiple flow voides with enlarged arteries and veins suggestive of arterio-venous malformations(AVMs)(Fig.2).

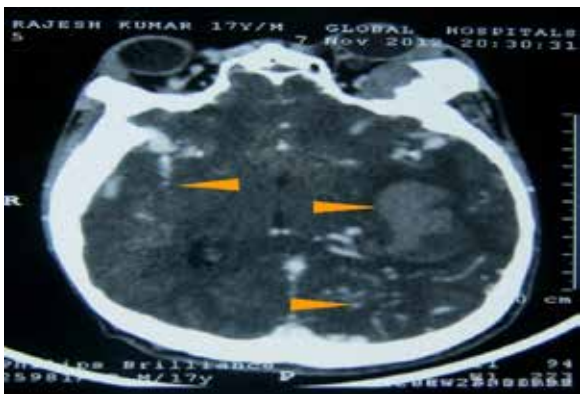

Fig2: MRI brain showing bleed in left temporal lobe and A-V Malformations

CT angio of brain revealed enlarged superior sagittal and right transverse sinuses; giant A-V malformations in brain parenchyma bilaterally(Fig.3 A & B); and bilateral proptosis with prominent intra-orbital veins.

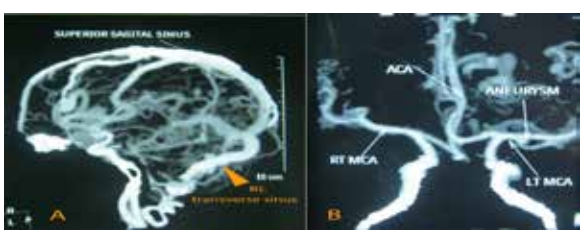

Fig3: CT Angio of brain showing dilated vessels in venous (A) and arterial (B) phases.

MRI spine showed cystic signal intensities suggestive of cystic angiomatosis; and Arnold chiari malformation type-I with tonsillar herniation(Fig.4 A & B).

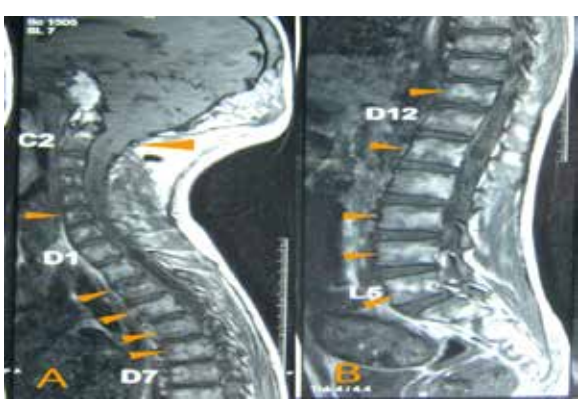

Fig4: MRI spine showing Arnold Chiari malformation (A) and multiple cystic lesions in vertebrae (B)

Biopsy was done, with sample taken from enlarged mandible. Histological examination revealed cystic spaces comprising RBC in between trabeculae and cystic spaces were lined by a single layer of endothelium, confirmative of skeletal cystic angiomatosis (Fig.5).

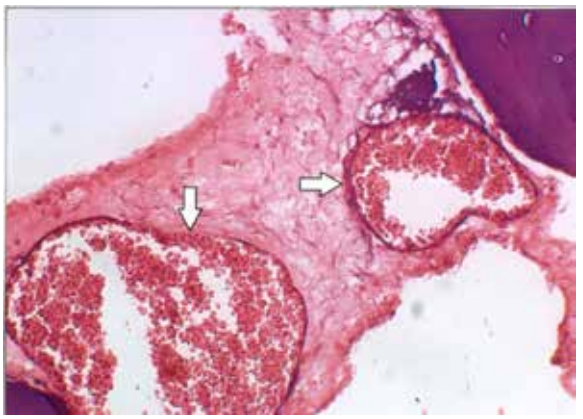

Fig5: Biopsy of Bone showing cystic spaces

So our patient had very rare congenital triad of abnormalities including skeletal cystic angiomatosis (CA) involving calvarium, facial bones and vertebrae; A-V malformations involving brain and tongue; and Arnold chiari malformation type-I.

During hospital stay patient was treated symptomatically, with supportive measures. Unfortunately we lost the patient suddenly, probable cause of death being recurrent massive intra-cerebral bleed.

Discussion

Cystic Angiomatosis (CA)

Pathogenesis of skeletal CA is still uncertain. One theory favours that the multiple, endothelial-lined cystic lesions are the result of vascular malformations of congenital origin, rather than representing the true neoplasm[3]. It is possible that nests of the primitive vascular channels persist and form the basis for the later diffuse, multi-centric appearance of the lesions in CA[4]. Another theory has advanced the possibility of agenesis of thyroid C cells that secrete calcitonin and their agenesis could induce osteolysis[5].

CA usually presents during first, second, or third decades of life, although the disorder has been diagnosed incidentally in elderly patients[6]. 12 cases of skeletal CA are presented in a family over four generations[7]. Inheritance pattern appears to be autosomal dominant with equal sex distribution. CA may be confined to the skeleton, as happened in our case, or may be associated with extensive soft tissue involvement, in which case spleen almost always is affected. Clinical features depend on the site of involvement.

Pathologic diagnosis of CA should be made in association with typical radiographic findings and clinical features. Microscopically contain numerous, dilated, cavernous thin walled vascular channels that are lined by flat endothelial cells[6]. The distinction between a hemangioma and a lymphangioma may be difficult, particularly if the cystic spaces are filled with fluid.

Lesions of CA are lytic, well defined, round or oval within the medullary cavity and they have intact cortex[8]. The lesions manifest variable peripheral sclerosis and exhibit endosteal scalloping. Periosteal reaction is typically absent[9]. Involvement of the axial and proximal appendicular skeleton is most common. Bones involved in decreasing order of frequency are femur, ribs, vertebrae, skull, innominate bone, humerus, scapula, tibia, radius, fibula and clavicle.

Differential diagnoses(D/D) of Skeletal C A[10]

The main D/D are A) In multiple myeloma the lesion tends to be similar in size and age of onset is 60-70 years. Anaemia, protienuria and increased monoclonal IG levels are the features. B) In skeletal metastases, a primary source may be identified and stability of bone lytic lesions without fracture for more than one year is unusual. C) Eosinophilic granuloma is typically unifocal, but may be multifocal and may show peri-

osteal reaction. D) Polyostotic fibrous dysplasia is associated with unilateral lesion with ground glass appearance, and abnormal skin pigmentation. E) Enchondroma typically shows calcifications. F) Gorhams disease is an aggressive form of skeletal angiomatosis, frequently involving shoulder and hip areas and usually monostotic.

Treatment Of Skeletal C A

There is no specific treatment for the bony lesions of CA. Local radiation and chemotherapy have been used in progressive cases, with no consistent change in the clinical course of disease. Corrective orthopaedic procedures may be indicated for pathological fractures. Because CA is invariably benign, amputation is rare, if ever is performed[6].

Prognosis of CA

Typical course of CA is relatively benign, not in-compatible with a long life span. When the disease involve viscera, dramatic clinical abnormalities may be evident, that contribute to the patient's early demise, as happened in our patient, in whom there were associated potentially dangerous malformations[6].

A-V Malformations of Brain

Our patient had intra-cerebral as well as lingual arterio-venous malformations(AVMs). AVM is a direct communication between one or more arteries and one or more draining veins without the intervening capillary bed. AVMs may occur anywhere in the intracranial spaces and may appear as isolated pathology or associated with other diseases. Most serious complication is intra-cerebral haemorrhage which occurs in 60% cases. 10% die with first haemorrhage and another 10% die of second haemorrhage, which was probable cause in our patient[11]. Treatment of AVMs include surgical resection or embolisation. Small AVMs may be obliterated by radio-surgery.

ARNOLD-CHIARI Malformation

Our patient had Arnold-chiari malformation type-I which is defined as an extension of the cerebellar tonsil, below the foramen magnum, for at-least 3-5mm. Chiari malformation may be associated with syringomyelia. Our patient had no symptoms of chiari malformation or of syringomyelia. Surgical decompression is needed in chiari malformation, in the presence of symptoms, usually type-II [11].

Conclusion

Our patient had an unusual combination triad of congenital malformations, which is rare and could not be identified in literature so far. CA is relatively compatible with prolonged life and documented as benign. Chiari mal-formation type-I was asymptomatic in our patient. Most potentially dangerous association was AVMs of brain, which was the probable cause of our patient's demise, in the form of intra-cerebral haemorrhage.

Acknowledgements

We thank Dr R V Subramanyam, Principal, Professor & Head, Oral Pathology,

Drs Sudha & Nageswara Rao Siddhartha Institute of Dental Sciences Chinoutapally, Gannavaram, A.P, India, for providing Histopathology slides in our case.

REFERENCES

1. Marco Pavanello, Gianluca Piatelli, Marcella Ravegnani,Alessandro consales, Andrea Rossi, Paolo Nezza, et al. Cystic Angiomatosis of the Craniocervical junction associated with chiari I malformation. Childs Nerv syst 2007; 23:697-700. | 2. Mirra JM, Arnold WD. Skeletal hemangiomatosis in association with hereditary hemorrhagic telangectasia. A case report. J Bone Joint Surg [Am] 1973; 55 : 850. | 3. Reid AB, Reid IL, Johnson G, Hamonic M, Major P. Familial Diffuse cystic angiomatosis of bone. Clin Orthop Rel Res 1989; 238:211-18. | 4. Boyle WJ. Cystic angiomatosis of bone. A report of three cases and review of the literature. J Bone Joint Surg Br 1972; 54:626-36. | 5. Korsic M, Jelasic D, Potocki K, Giljevic Z, Aganovic I. Massive osteolysis in a girl, with agenesis of thyroid C Cells. Skeletal Radiol 1998;27:525-8. | 6. David S.Levey, Lisa M.Mac cormack, David J.Sartoris, Parviz Haghighi, Donald Resnick, Roger Thorne. Cystic Angiomatosis: case report and review of the literature. Skeletal Radiol 1996; 25:287-93. | 7. Reid AB, Reid IL, Johnson G, Hamonic M, Major P. Familial diffuse cystic angiomatosis of bone. Can J surg 1987; 30(4):277-9. | 8. Lomasney LM, Martinez S, Demos TC, Harrelson JM. Multifocal vascular lesions of bone: imaging characteristics. Skeletal Radiol 1996; 25:255-61. | 9. Lateur L, Simeons CJ, Gryspeerdt S, Samson I, Mertens V, Van Damme B. Skeletal cystic angiomatosis. Skeletal Radiol 1996; 25:92-95. | 10. Tedric D.Boyse, MD, Jon A.Jacobson MD. Cystic Angiomatosis Radiology 2002; 223: 164-67. | 11. Janet Eyre. Neurodevelopmental disorders. In:Michael Donaghy, editor. Brains Diseases of Nervous system.12th ed.Newyork: Oxford University press Inc; 2009.p.221-25.